

Supplemental Data

A Missense Mutation in SLC33A1,

which Encodes the Acetyl-CoA Transporter,

Causes Autosomal-Dominant Spastic Paraplegia (SPG42)

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Table S1 Two-Point LOD Scores Showing Exclusion of Linkage to Known HSP Loci

Candidate Gene	Chr.	Marker	LOD Score ($\theta = 0$)*
SPG3A	14q11-q21	D14S589	-7.273
SPG4	2p22-p21	D2S2351	-11.111
		D15S817	-8.814
SPG6	15q11.1	D15S128	-8.990
		D15S210	-9.291
		D15S646	-8.990
SPG8	8q24.13	D8S1179	-13.115
		D12S390	-16.464
SPG10	12q13	D12S1691	-16.990
		D12S298	-16.405
SPG12	19q13	D19S587	-9.914
		D2S425	-4.333
SPG13	2q33.1	D2S2318	-4.530
		D2S1384	-4.781
SPG19	9q33-q34	D9S934	-13.352
SPG29	1p31.3-p21.1	D1S1588	-9.944
SPG31	2p11.2	D2S428	-3.222
		D2S2951	-5.631
SPG33	10q24.2	D10S677	-5.893

* A disease allele frequency of 0.0001 and a penetrance of 90% were assumed for the disease locus.

Table S2. Linkage-Scan Results

Chr.	No. of Genotyped Markers	Intermarker Distance (cM) Range (Average)	Maximum LOD Score	No. of Markers with LOD Scores > -2	Markers with Positive LOD Scores
1	16	10.25-23.96 (18.1)	-3.437	0	
2	18	7.05-21.7 (15.0)	-3.148	0	
3	14	6.42-22.18 (16.3)	3.569	2	D3S1744: 3.569; D3S3053: 0.797
4	13	9.63-20.56 (16.3)	1.605	1	D4S2431: 1.605
5	13	9.09-22.35 (15.2)	-8.26	0	
6	11	8.53-22.35 (17.6)	1.566	1	D6S1959: 1.566
7	11	6.49-22.75 (16.5)	0.362	1	D7S517: 0.362
8	11	6.92-22.41 (15.3)	-6.907	0	
9	10	11.32-22.76 (16.9)	-3.365	0	
10	11	11.49-21.74 (15.7)	-3.419	0	
11	8	15.08-25.97 (18.5)	-7.248	0	
12	10	6.42-23.35 (17.6)	-3.192	0	
13	7	8.87-23.94 (16.3)	-5.36	0	
14	9	10.18-21.37 (15.4)	-6.55	0	
16	7	12.9-22.65 (19.2)	-6.951	0	
17	7	12.55-23.52 (18.1)	-9.497	0	
18	7	12.81-23.24 (18.0)	-6.437	0	
19	7	7.48-22.53 (15.0)	-6.054	0	
20	6	9.53-22.07 (16.9)	0.331	1	D20S162: 0.331
21	4	13.05-17.28 (14.4)	-6.089	0	
22	5	4.06-16.49 (12.5)	-2.912	0	
Total	205			6	

Table S3. Primers Used for PCR Amplification of *SLC33A1* Exons

Exon	Primer (5'→3')		Product Size (bp)	Annealing Temperature (°C)
	Forward	Reverse		
1	GTGCCCTTATCGCTCTGA	GTGTTATTTGATGGGTTGC	1155	55
2	CCTGCCTTTTTGGTTGTTGT	TGGTAGTGGAAAGGGACCTG	818	63
3	TGACTCATGGGGAGATACT	AACTCAGCAGGACTACGAT	1176	54
4	ATTGGTAGAAGAGGGAGTAC	TTAAGACAACCTGTAGAAAGC	615	58
5	GTCACATCTTTGTATTGCTGG	ACGCCTTTAAGGGAAGTAG	954	57
6	CACCCGTAATTCCAGCATT	CACCCGTGACTACTGCATTG	976	63